Congenital Skeletal Dysplasia

Skeletal dysplasias are a heterogeneous group of more than 200 disorders characterized by abnormalities of cartilage and bone growth resulting in abnormal shape and size of the skeleton and disproportion of the long bones, spine, and head. In general, patients with disproportionately short stature have skeletal dysplasia (osteochondrodysplasia). Dwarfism is a commonly used term for disproportionately short stature, although a more medically appropriate term for this disorder is skeletal dysplasia. Short stature is defined as height that is 3 or more standard deviations below the mean height for age. If short stature is proportional, the condition may be due to endocrine or metabolic disorders or chromosomal or nonskeletal dysplasia genetic defects.

A very small number of these disorders are not compatible with life and infants will die soon after birth. Skeletal dysplasias include the following disorders: achondrogenesis, homozygous achondroplasia, chondrodysplasia punctata (recessive form), camptomelic dysplasia, congenital lethal hypophosphatasia, perinatal, lethal type of osteogenesis imperfecta, thanatophoric dysplasia, and short-rib polydactyly syndromes.

Overall incidence of skeletal dysplasias is approximately 1 case per 4,000 to 5,000 births. It has been suggested that the true incidence may be twice as high because many skeletal dysplasias do not manifest until short stature, joint symptoms, or other complications arise during childhood.

Lethal skeletal dysplasias are estimated to occur in 0.95 per 10,000 deliveries. The three most common skeletal dysplasias are *thanatophoric dysplasia, osteogenesis imperfecta*, and *achondrogenesis*. Thanatophoric dysplasia and achondrogenesis account for 62% of all lethal skeletal dysplasias.

Achondrogenesis is dwarfism accompanied by various bone aplasias of all four limbs, a normal or enlarged cranium, and a short trunk with delayed ossification of the lower vertebral column and pelvic bones. Lethal achondrogenesis types I and II are both rare. Their respective frequencies are not known; however, the overall frequency has been estimated at 1 in 40,000 births.

Thanatophoric dysplasia (TD) is the most common form of lethal skeletal dysplasia in the neonatal period. Complications include the following: severe growth and developmental delay, hydronephrosis, hydrocephalus, and ventilator dependency. The frequency of TD is 1 per 10,000-35,000 live births. TD is usually lethal in the first few days of life. Death is caused by respiratory insufficiency.

Osteogenesis imperfecta (OI) is an abnormal fragility and plasticity of bone, with recurring fractures on trivial trauma; variable associated features include

deformity of long bones, blueness of sclerae, laxity of ligaments, and otosclerosis. It is also referred to as brittle bones.

The prevalence of OI is estimated to be 1 per 20,000 live births, but the mild form is underdiagnosed and the actual prevalence may be higher than this.

NOTE: This Web page was compiled from a variety of sources including the online resources of eMedicine, Nemours, St. Louis Children's Hospital, and other resources listed below, but is not intended to substitute or replace the professional medical advice you receive from your physician. The content provided here is for informational purposes only, and was not designed to diagnose or treat a health problem or disease. Please consult your health care provider with any questions or concerns you may have regarding this specific condition.

RESOURCES

NOTE: This page contains links to other World Wide Web sites with information about this disorder. The Department of Health and Senior Services (DHSS) hopes you find these sites helpful, but please remember the DHSS does not control nor does it necessarily endorse the information presented on these Web sites.

For a complete list of resources related to birth defects, including state programs and resources, support groups and not-for-profit organizations click on the following link. http://www.dhss.mo.gov/BirthDefects/Resources.pdf

eMedicine from Web MD
http://www.emedicine.com/

Genetic Alliance
http://www.geneticalliance.org

Nemours
Http://www.Nemours.org/

St. Louis Children's Hospital
http://www.stlouischildrens.org.

GENETIC TERTIARY CENTERS

How to Obtain Genetic Services
Your family physician can usually provide information regarding genetic services

in your area. Genetic clinics are periodically held in a location near you. For information please contact one of the centers listed below.

Cardinal Glennon Children's Hospital
St. Louis, Missouri
314-577-5639

Children's Hospital at University Hospital and Clinics
Columbia, Missouri
573-882-6991

Children's Mercy Hospital
Kansas City, Missouri
816-234-3290

St. Louis Children's Hospital
St. Louis, Missouri
314-454-6093